What is a Genetic Counselor?
Genetic counselors are specialists who provide personalized help about your genetic health.
What is a Genetic Counselor?
Genetic counselors work in a variety of settings.
The largest specialty areas for genetic counselors are cancer, prenatal and pediatric genetics, but genetic counselors work in a variety of specialties.
What do genetic counselors do?

- Help you understand your genetic risks based on your family history
- Help develop a personalized medical plan
- Enable you to make an informed decision about testing
- Explain results and help you process the medical emotional implications
- Connect you to resources, support groups, other patients
Reasons to See a Genetic Counselor

• You are pregnant or considering pregnancy and have questions about your baby inheriting a condition

• You, your child or a family member has been diagnosed with a genetic condition, or there is concern for a genetic condition

• Your or a family member have unexplained medical concerns that could be due to genetic causes
Common Diseases that May Have a Genetic Component

- Cancer
- Heart disease
- Diabetes
- Hypertension
- Stroke
- Alzheimer's
- Arthritis
- Osteoporosis
Preparing for your visit with a Genetic Counselor

- Family health history
- Medical records
- Results of any genetic tests
- A list of your questions and concerns
What happens during a genetic counseling appointment?

- Genetic counselors ask about your family health history, discuss genetic risks and testing options and support you.
- Talk about any appropriate options in terms of genetic testing and medical management.
- Help you understand available tests and possible results, and help you decide when and if to pursue genetic testing.
How much does genetic counseling cost?

- Health insurance typically pays for genetic counseling and in many cases pays for genetic testing when it is recommended by a doctor.
- Check with your insurance company to verify coverage.
Explore a Career as a Genetic Counselor
There are so many reasons to become a Genetic Counselor.
Genetic Counselor was named one of the “25 Amazing Healthcare Support Jobs” and one of the “100 Best Jobs” in 2017.

- U.S. News and World Report
By the Numbers...

Employment of genetic counselors is projected to grow 29% from 2014-2024. Average growth is 7%.
- Bureau of Labor Statistics

The average salary for a full-time genetic counselor in 2015 was $81,377 and ranges up to $250,000 depending on specialty area and experience.
- NSGC 2016 Professional Status Survey

The job outlook for genetic counselors is excellent.
- ExploreHealthCareers.org

90% of genetic counselors report they are satisfied with their job
- NSGC 2016 Professional Status Survey
The Education Path

A bachelor’s degree, typically in:
• biology
• social science
• or a related field

A master’s degree in genetic counseling from an accredited program:
• coursework
• clinical training
• research

ABGC and/or ABGCC Certification (USA & Canada) after passing the Board Examination. Many states also require licensure to practice.
Genetic Counseling Program Coursework

- Human genetics
- Embryology
- Ethics
- Counseling techniques
- Genetic tests
Interested in Applying to a Genetic Counseling Program?

For tips and information, visit the Association of Genetic Counseling Program Directors

https://agcpd.org/AdmissionsInfo
A Range of Career Possibilities

- Working with patients
- Laboratories
- Research
- Academia
- Professional and patient organizations
- Healthcare administration
- Public health
- Policy
A Range of Career Possibilities

Any role you can imagine and create for yourself

“As a genetic counselor, I've had the opportunity to work in many different areas of medicine. And the impact I've had over the course of those years…. has been as an advocate for patients.”

-Janet Williams, CGC
Why and when should I refer to a Genetic Counselor?

“Working with a genetic counselor can fill in the gaps between offering genetic testing and making sure a patient receives appropriate discussion about testing, implications of genetic testing and when it is necessary [to help test] other family members.”

— Doctor, Vascular Medicine
Why work with a Genetic Counselor?

• Increase patient satisfaction
• Make informed decisions
• Save time and money

“I learned a lot myself from the genetic counselor progress notes about next steps in treatment and workup, and my patients are reassured by having greater understanding of the test results.”

— Doctor, Family Practice
Prenatal Indications for Genetic Counseling

- Mother will be 35 years or older at time of delivery (singleton)
- Mother will be 33 years or older at time of delivery (twins)
- Consanguineous union
- Abnormal first or second trimester maternal screening
- Exposure to known or potential teratogen
- Fetal anomaly/anomalies
- Positive carrier screen for genetic condition
- Personal history of stillbirths, previous child with hydrops or SIDS, recurrent pregnancy losses
- Either partner with a family history of birth defects, chromosomal anomalies, potentially genetic hearing/vision loss, intellectual disability or autism
Oncological (Cancer) Indications for Genetic Counseling

- Personal or family history of cancer(s) known to be associated with specific genes or mutations, such as:
  - Breast
  - Ovarian
  - Colorectal
  - Uterine
  - Pancreatic
  - Gastric
  - Neuroendocrine
  - Metastatic prostate cancer

- This should be considered in the context of:
  - A compelling family history
  - Young age at onset
  - Bilateral lesions
  - And/or familial clustering of related tumors

- Any child with a solid tumor
Pediatric Indications for Genetic Counseling - Newborn/Infant

- Abnormal newborn screening results
- Born to a parent with a known chromosomal abnormality or rearrangement (balanced or unbalanced)
- A single major, or multiple major and/or minor anomalies
- Dysmorphic features
- Hypotonia or hypertonia
- Failure to thrive

- A known metabolic disorder or symptoms of a metabolic disorder (e.g., intractable seizures, hepatosplenomegaly, acidosis, cyclic vomiting, persistent hypoglycemia, developmental regression, and unusual body odor)
- Abnormal brain MRI findings (e.g., leukodystrophy, periventricular calcifications, unidentified bright objects, or a malformation)
Pediatric Indications for Genetic Counseling - Childhood

- Autism or pervasive developmental disorder
- Unusual behaviors, especially when associated with minor malformations and developmental delay
- Unexplained intellectual disability or global developmental disorder
- An unusual growth pattern such as overgrowth, short stature, or hemihypertrophy
- Evidence of a connective tissue disorder
  - (e.g., extreme joint laxity, poor wound healing, or a marfanoid habitus)
- Congenital eye defects or blindness associated with problems
  - (e.g., microphthalmia, cataracts, megalocornea, retinitis pigmentosa, or cone-rod dystrophy)
Pediatric/Adult Indications for Genetic Counseling

• A recognized or suspected genetic syndrome, including a chromosomal or single gene disorder
• A significant family history of medical or psychiatric conditions that puts the patient at risk of developing the same or similar condition
• Unusual skin findings (e.g., multiple types of lesions, multiple lipomas, numerous hypo or hyperpigmented lesions, and albinism)
• Problems with clotting, including disorders such as hemophilia and thrombophilia
Pediatric/Adult Indications for Genetic Counseling

- Progressive muscle weakness that might be associated with a genetic disorder (e.g., a form of muscular dystrophy, spinal muscular atrophy, or myotonic dystrophy)
- Other neurologic condition that might be associated with a genetic predisposition (e.g., peripheral neuropathy, unexplained myopathy, progressive ataxia, or any progressive neurologic disorder without a clear, nongenetic cause)
- An immunodeficiency or significant immune problem
- Cardiomyopathy not secondary to a viral infection
Pedigree: also called a “family tree”; a tool genetic counselors use to visualize a family health history and identify recognizable patterns.
Cancer Genetics
Meet Mary

• 45-year-old woman recently diagnosed with breast cancer

• Family history of cancer, including a mother with ovarian cancer who passed away at 70

• Especially concerned about risks for her daughters (ages 22 and 25)
Mary’s Family History

- The diagram shows a family tree with symbols indicating individuals and their ages.
- Red dots indicate breast cancer, and blue dots indicate ovarian cancer.
- The ages of individuals are marked: 75, 45, 25, and 22.
- The diagram illustrates familial relationships and medical history.
Hereditary Breast Cancers

- Around 10-15% of all breast cancers are hereditary.
- People with alterations in the genes *BRCA1* or *BRCA2* have greatly elevated chances to develop certain cancers, especially female breast (up to 85% risk) and ovarian (up to 45% risk).
- Other features may include:
  - Multiple affected individuals
  - Younger age of onset
  - Affected individuals may have:
    - bilateral breast cancer and/or triple negative breast cancer
    - multiple primary cancers which may include breast, ovarian, male breast, pancreatic, prostate, or melanoma cancers
In Mary’s Case

• She elected to undergo genetic testing

• A BRCA1 alteration was identified, confirming a diagnosis of hereditary breast and ovarian cancer syndrome (HBOC)

• Changed Mary’s medical management, including more screening and options for surgery and medications to reduce risks

• Allowed for testing of at-risk family members and early intervention in those with the same pathogenic variant
Meet Joe

- 45-year-old male recently diagnosed with colon cancer
- Family history of cancer, including a mother with uterine cancer who passed away at 50, a sister who had precancerous colon polyps, and a maternal uncle who had colon cancer at age 47
- Especially concerned about risks for his daughters (ages 22 and 25)
Joe’s Family History

- Colon Ca. 45
- Colon Polyps
- Uterine Ca. 49
- Colon Ca. 47
- d. 50
- 73

Symbols:
- Green square: Colorectal Cancer
- Yellow square: Uterine Cancer

Individuals:
- 45
- 43
- 25
- 22
Hereditary Colorectal Cancers

- Around 10% of all colorectal cancers are hereditary
  - Of those, the majority are due to Lynch syndrome caused by alterations in the MLH1, MSH2, MSH6, PMS2, or EPCAM genes.
- People with Lynch syndrome have greatly elevated chances to develop certain cancers, especially colorectal, uterine, ovarian, gastric, and a few others.
- Other features may include:
  - Younger age of onset
  - Precancerous colon polyps
- Individuals at high risk for colon cancer can significantly decrease this risk by having more frequent colonoscopies, beginning at a younger age.
In Joe’s Case

- Joe elected to undergo genetic testing
- An alteration in \( MSH2 \) was identified, confirming a diagnosis of Lynch syndrome
- Changed Joe’s medical management, including more screening and options for surgery
- Allowed for testing for at-risk family members and early intervention in those who also inherited the \( MSH2 \) alteration
Pediatric Genetics
4-year-old boy, David, comes to clinic due to short stature, developmental delay and an insatiable appetite.

Parents remark that he was very “floppy” as a baby.

The geneticist and genetic counselor immediately suspect a diagnosis, and counsel the family about testing options.
Prader-Willi Syndrome

- Rare genetic condition affecting ~1/30,000 people
- Majority of cases are sporadic
- Recommended treatment includes growth hormone therapy
- Very specific genetic testing indicated, which may not be understood by a healthcare practitioner outside of genetics
- Parents elect testing, which returns positive and David is diagnosed with Prader-Willi syndrome
Prenatal Genetics
Meet Ann

- 39-year-old woman, 13 weeks pregnant

Ann underwent prenatal screening through her OB/Gyn and was referred to genetic counselor to discuss her screening results.
During the Appointment

- Ann’s genetic counselor explained that her prenatal screening results indicated that her pregnancy had a higher chance of being affected with Down syndrome
  - Test revealed a 1/50 chance

- Ann discussed these results with her genetic counselor as well as possible next steps, which could include a diagnostic procedure (such as amniocentesis) for a prenatal diagnosis
• After meeting with her genetic counselor, Ann decided to undergo an amnioscentesis, a diagnostic test, which indicated a high risk for trisomy 21, or Down syndrome, in her pregnancy.

• Ann can use the information from the amnioscentesis to discuss with her doctor whether she should elect to deliver at a high-risk hospital and to prepare herself and her family members for the arrival of the baby.
Meet William

• A 50-year-old man comes to clinic after an echocardiogram revealed he had an enlarged aorta

• He had heard that some type of heart condition affected his relatives, but he hadn’t realized he could be at risk since he felt healthy

• Now, he would like to learn more about his personal and family’s risks
William’s Initial Family History

Unknown cause of death

- 50: Aortic dilation
- 55
- 21
- 19
- 61
- 42
- 48: Heart surgery, age 45, Obese
- 28: Drowning
- 32: Pregnancy complication
William’s Family History After Genetic Counseling

Diagram showing family members and their medical conditions:
- d. 60 with Aortic dissection
- 51
- 55 with Aortic dilation
- 50 with Aortic dilation
- d. 28 with Drowning
- Heart surgery, age 45
- 48
- d. 32 with Aortic aneurysm during pregnancy
- 42
- 21
- 19

Legend: □ Aortic dilation

Genetic Counselor Awareness Day
Partners in Your Genetic Health Care
#IAmAGeneticCounselor
During the Appointment

• We learned that his two deceased siblings had both passed away due to major medical problems with their aortas

• These events had been explained away due to pregnancy or drowning, so the emerging pattern was not immediately clear

• William was offered genetic testing for heritable thoracic aortic disease
Moving Forward

- William can work with his genetic counselor and family to determine whether he wants to pursue testing.

- A positive test could allow his children to obtain testing themselves and obtain proper screening and medical care.
General Genetics
Meet Gianna

- A 28-year-old woman, Gianna, comes to General Genetics clinic, with a history of chronic pain
- She had been to many other doctors’ appointments, and had been told that her symptoms were likely psychiatric
- Gianna also has a history of depression and anxiety
- Many “chronic pain” cases do not have a genetic explanation, however
During the appointment...

- Genetic counselor asks for full, detailed history of pain, and Gianna explains she has severe stomach pain, and she often feels like hands and feet are burning or tingling

- The genetic counselor reviews medical records, and notices that in a routine urine test, Gianna was noted to have protein in her urine

- Gianna was adopted, and does not know any health information about her biological family
Gianna’s Diagnosis

• Gianna is offered testing for Fabry disease, which confirms her diagnosis

• This condition has treatment, which helps Gianna’s symptoms improve

• Her diagnostic information can also be used for family planning, if Gianna is considering pregnancy
Moving Forward

• Gianna’s genetic counselor gives her a flyer for a national Fabry support group, which she joins and meets other people with the same condition

• She is started on enzyme replacement therapy infusions every other week, which improves her symptoms

• Gianna is grateful that her genetic counselor took her concerns seriously, listened to her story and advocated for her
Laboratory
In a Genetic Testing Laboratory

- Phone call from a physician at a high-risk breast cancer clinic
- Patient is 52-year-old woman, who has never had cancer
- She had genetic testing 4 years ago at another doctor’s office, which revealed a variant of uncertain significance (VUS) in a gene called BRCA1
In a Genetic Testing Laboratory

- Family history: Patient’s mother was diagnosed with breast cancer at 55, sister diagnosed with breast cancer at 45

- Physician calls the lab to speak with a genetic counselor, and asks if additional genetic testing is necessary
During the Phone Call

• Laboratory genetic counselor notes that a previously-reported variant of unknown significance (VUS) has now been reclassified as a “pathogenic variant”

• There are established medical guidelines to guide medical management, such as additional surveillance or surgery

• In addition, now specific testing may be offered to other, at-risk family members
Genetic Counselor Awareness Day
Partners in Your Genetic Health Care

#IAmAGeneticCounselor